Suggested Follow-up for Congenital Hypothyroidism Elevated TSH

Possible Causes: Congenital hypothyroidism is usually caused by total or partial failure of development of the thyroid gland (aplasia or hypoplasia) or development of the gland in an abnormal place (ectopic gland.) Newborn screening specimen includes measurement of both TSH and T4. The physiologic surge in TSH during the first hours of life can affect its measurement, causing false positive screening results in some infants, depending upon the timing of specimen collection.

Next Steps if Abnormal: Repeat T4/TSH screening on filter paper and send to the DHEC laboratory. No treatment necessary until result of repeat testing known. Consider serum thyroid panel and consult pediatric endocrinologist for further instructions if TSH is over $40 \,\mu\text{IU/mL}$.

Neonatal Presentation: Usually none.

Emergency Treatment: None.

Standard Treatment: Thyroxine replacement medication.

Advice for Family: Provide basic information about congenital hypothyroidism. The handout, When Baby Needs a Second Test for a Congenital Hypothyroidism (Elevated TSH), may be used for this purpose.

Internet Resources:

http://www.nlm.nih.gov/medlineplus/ency/article/001193.htm

http://www.emedicine.com/ped/topic501.htm

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm